Suggested Follow-up for Elevated C5DC: Glutaryl Carnitine

Possible Causes: Elevated C5DC is the primary marker for **glutaric aciduria type I (GA I).** It is a disorder of tryptophan (TRP), lysine (LYS) and hydoxylysine metabolism.

Next Steps if Abnormal: **Potential medical emergency.** See infant as soon as possible to ascertain health status. Collect blood spot specimen for repeat acyl carnitine profile. A portion of the initial specimen will be sent to the Greenwood Genetic Center Laboratory for secondary testing. Common diagnostic studies include plasma total and free carnitines, plasma acylcarnitines and urine organic acids.

Neonatal Presentation: Macrocephaly, irritability, jitteriness, hypotonia, acidosis, ketosis, hypoglycemia and hyperammonemia. Infants are at risk for encephalopathic crisis and metabolic decompensation/crisis.

Emergency Treatment: Treatment of metabolic crisis includes provision of sufficient calories (concentrated dextrose infusion with appropriate electrolytes) to correct catabolic state and biochemical abnormalities if needed.

Standard Treatment: Prompt treatment of catabolic events. Aggressive fever control. Watch fluid intake, as profuse sweating may occur. Protein restricted diet with use of metabolic formula without LYS and TRP. Riboflavin trial. Carnitine supplementation.

Advice for Family: Provide basic information about organic acid disorders. The handout, *When Baby Needs a Second Test for an Organic Acid Disorder (Elevated C5DC)*, may be used for this purpose. Stress the importance of seeking immediate medical attention if the infant shows any signs of illness.

Internet Resources:

http://oregon.gov/DHS/ph/nbs/expand.shtml

http://web1.tch.harvard.edu/newenglandconsortium/scientists_physicians2.html

http://ghr.nlm.nih.gov/condition=glutaricacidemiatypei

http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm